Long-term outcome in open spina bifida

Pippa Oakeshott and Gillian M Hunt

SUMMARY

Background: *Doctors need reliable data on outcome in order to help parents faced with difficult decisions about termination of an affected pregnancy or treatment after birth.*

Aim: *To determine survival, health and lifestyle at the mean age of 30 years in a complete cohort of adults born with open spina bifida.*

Design of study: *Prospective cohort study.*

Participants: *Well-documented cohort of 117 consecutive cases of open spina bifida whose backs were closed non-selectively at birth between 1963 and 1971.*

Method: *Survivors (age range = 26 to 33 years) were surveyed by postal questionnaire and telephone interview. The main outcome measures were the health, independence and lifestyle of the survivors in terms of living in the community, driving a car and working in open employment.*

Results: *Ascertainment was 100%. Sixty (51%) had died, mainly the most disabled. Of the 57 survivors, 84% had a cerebrospinal fluid (CSF) shunt, 70% had an IQ of 80 or more, 37% lived independently in the community, 39% drove a car, 30% could walk more than 50 metres and 26% were in open employment. However one-third (19) still needed daily care, three were on respiratory support, two were blind, two had diabetes mellitus, and one was on dialysis. Mortality, disability and achievement reflected the neurological deficit that had been recorded in infancy in terms of sensory level. Attainment and independence were reduced in those who had needed revision of CSF shunt.*

Conclusion: *The survivors in this unselected cohort showed a wide range of outcome from apparent normality to very severe disability. This reflected both the extent of their original neurological deficit and events in the history of their CSF shunt.*

Keywords: *Open spina bifida; outcome; attainment; CSF shunt.*

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Introduction

IN the 1960s, when this study started, babies with open spina bifida who owed their lives to the cerebrospinal fluid (CSF) shunt were not expected to survive into adulthood. Their future disability was unknown, as were its implications. Although early results of treatment led to optimistic forecasts, their physical and intellectual defects became more apparent as they grew older, and reviews became less encouraging.[1-5] Data on long-term outcome of open spina bifida are vital for doctors helping parents with difficult decisions about termination of an affected pregnancy or treatment after birth. In the UK, most adults with spina bifida are cared for by general practitioners (GPs). The aims of this review were to record survival, health, lifestyle and attainments at the mean age of 30 years in a complete cohort of adults with spina bifida.

Method

Patients

In 1963 paediatricians in East Anglia agreed to refer all newborn babies with spina bifida to the Regional Neurosurgical Unit at Addenbrooke's Hospital, Cambridge. Treatment was offered to all babies with open spina bifida without any attempt at selection, provided they were seen within a few hours of birth. Between 1963 and 1971, after a full neurological examination, 117 babies (50 male, 67 female) had their defects closed within 48 hours of birth. A CSF shunt was inserted for hydrocephalus when required.

Data collection

In 1997 all survivors were surveyed by confidential questionnaire and telephone interview. They were asked about their health, disability and attainments in terms of living independently, driving a car and working in open employment. For those who had died, causes of death were obtained from medical records and the Office of National Statistics. The study was approved by Cambridge Local Research Ethics Committee.

Statistical analysis

Patients had previously been classified into four groups according to sensory level to pin prick recorded in infancy.[4] Those with intact sensation down to the knee (sensory level below L3) had a better short-term outcome than those with intact sensation no lower than the umbilicus (sensory level above T11). Mortality and measures of disability and attainment were compared in those with different sensory levels and CSF shunt histories using χ^2 for trend.

Results

Ascertainment was 100%. Twenty-six (46%) of the 57 survivors responded to the questionnaire, and all survivors or a carer or relative were interviewed by telephone.

HOW THIS FITS IN

What do we know?

The impact of treatment in open spina bifida has been highly successful in reducing mortality, but its influence on disability is often overestimated and short-lived. Doctors helping parents faced with difficult decisions about termination of an affected pregnancy or treatment after birth need reliable data on outcome.

What does this paper add?

This is the first prospective study of a complete and unselected series of patients with open spina bifida followed up for 30 years by the same observer. By the mean age of 30 years the majority of the most severely affected cases had died. Although a third of the survivors continued to need daily help, a further third lived independently. Outcome was related both to the neurological deficit in infancy and to the history of the CSF shunt. The best outcome was seen in those with a low sensory level who had no CSF shunt, or in those with a shunt who had remained free of symptomatic episodes of shunt insufficiency. Since lack of attainment and blindness could follow symptomatic shunt insufficiency, general practitioners need to recognise and refer promptly any patient suspected of having raised intracranial pressure.

Mortality

Figure 1 summarises the outcome, including causes of death, for the complete cohort. Sixty patients (51%) had died: 25 before their first birthday and a further 15 before their fifth birthday. Thereafter, the death rate remained constant with an average of 1% of the remainder dying each year.[6] Those with a high sensory level above T11 had an increased mortality over those with a low sensory level below L3 (29/42 versus 13/38, relative risk [RR] = 2.0, 95% confidence interval [CI] = 1.2 to 3.2). This difference was mainly owing to the increased mortality from renal causes in those with a high sensory level (14/42 versus 0/38, $P = 0.001$).

Survivors

There were 57 survivors: 25 male, 31 female and one who had undergone a sex change from male to female. The mean age was 30 years (range = 26 to 33 years). The disability of the survivors ranged from a blind, paraplegic, doubly incontinent woman who needed total care, to an intelligent, athletic business manager whose only stigmata were a scar on his back and a shunt in his head. Table 1 shows that sensory level in infancy was a predictor of disability, the need for a CSF shunt, IQ, need for a wheelchair or daily care, and attainment.

CSF shunts

Of the 57 survivors, nine had never had a shunt; of these, eight had little or no disability and a sensory level below L3. The remaining 48 had had a ventriculo-atrial shunt inserted. In 16 patients the shunt had never been revised. The other 32 patients had had a total of 113 revisions: for shunt insufficiency (60%), infection (16%), detachment (13%), extrusion or leaking wound of back (5%), and unknown (6%). In ten patients revisions were done only before the age of two years (mean = 1.3 revisions, range = 1 to 3), and in 22 patients revisions were carried out between the ages of two and 30 years (mean = 4.5 revisions, range = 1 to 14). Elective revisions were not performed; and shunts were inserted or revised only in response to definite clinical need. Of those who had revisions, 75% had had symptoms of raised intracranial pressure.

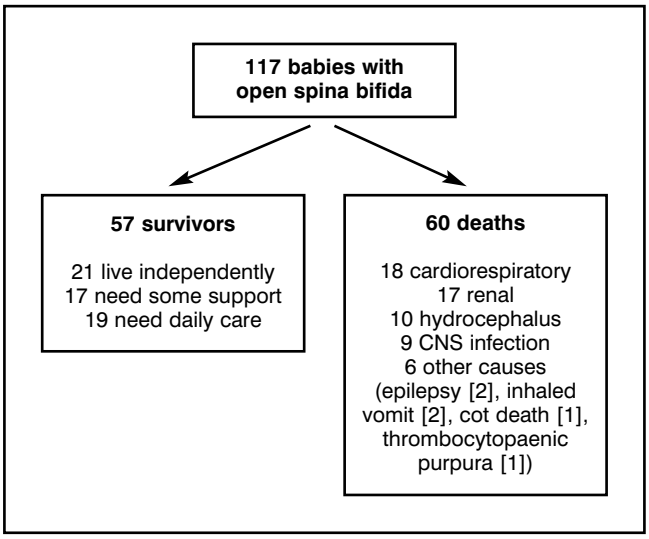

Figure 1. Outcome in open spina bifida at the mean age of 30 years.

Intelligence

Forty of the 57 survivors (70%) were of normal intelligence (IQ greater than 80); these included the nine who had never needed a shunt. Two survivors, both of whom were of normal intelligence when assessed at school,[7] later suffered brain damage and loss of vision associated with shunt failure and prolonged apnoea, and were reassessed as having an IQ lower than 80.

Mobility

Only 17 (30%) remained community walkers, defined as being able to walk more than 50 metres with or without aids. Ten of the 17 could walk at least a kilometre. Table 2 shows the deterioration in walking since childhood and its relationship to sensory level. By the age of 30 years, there were no community walkers with a sensory level of L3 and above. But of those with a sensory level of L5 and below, 88% (15/17) remained walkers. In terms of motor function, 31 had been recorded as having bilateral quadriceps activity in infancy, but only 55% of them (17/31) remained walkers.

Health

During the previous five years, nearly half of the survivors had been in hospital. The main reasons were urological (11 patients), shunt dysfunction (four patients), pressure sores (three patients), and sepsis (three patients). One patient was on dialysis and three had nocturnal respiratory support. Two individuals were totally blind following shunt dysfunction and four others had severe visual defects. Endocrine conditions were common: two patients had diabetes mellitus, one had adrenal hyperplasia, one had primary azoospermia, and six had had precocious puberty (defined as menstruation [three] or signs of puberty in the male [three] before the tenth

Table 1. Sensory level in infancy related to disability at mean age 30 years in 57 survivors with spina bifida.

Sensory level (n = 57)	n (%)	Below L3 (n = 25)	L3–T11 (n = 16)	Above T11 (n = 13)	Asymmetrical (n = 3)	χ^2 for trend[a]
Severe disability[b]	21 (37)	0	8	13	0	$P<0.0001$
CSF shunt	48 (84)	17	16	12	3	$P<0.05$
IQ <80	17 (30)	3	5	7	2	$P<0.01$
Wheelchair	40 (70)	9	16	13	2	$P<0.0001$
Daily care needed	19 (33)	4	5	10	0	$P<0.001$
No attainment[c]	29 (51)	7	10	10	2	$P<0.01$

[a]Patients with lower sensory levels have less disability. Asymmetrical sensory level excluded from the analysis. [b]Severe disability defined as wheelchair-dependent, needs help with transfers, continence care and daily living, mostly low IQ, kyphosis, pressure sores, epilepsy and visual defects (two blind). [c]No attainment in terms of living independently, driving a car or working in open employment.

birthday). Twenty-six patients were on long-term therapy: anticonvulsants (11 patients), antibacterials (10 patients), and antihypertensive drugs (nine patients). Six further patients had needed long-term anticonvulsants in the past, giving an overall incidence of epilepsy of 30%. Four patients needed regular analgesics for musculo-skeletal pain.

Parenthood

Seven women and one man had become parents. The man had minimal disability and no detectable sensory loss.

Residence and dependency

Twenty-one individuals (37%) lived independently in the community, ten of whom used wheelchairs. A further 17 (30%) were personally independent but had supervision and help when required. The remaining 19 (33%) needed help daily for dressing, shaving, toilet, or nursing care (mainly pressure sores). Thirteen of these still lived with their parents who were now aged 47 to 77 years; two women were in the care of their husbands; three were in residential establishments; and one was in sheltered accommodation with 24-hour attendance.

Car drivers

Thirty-one (54%) of the survivors had passed their driving test, but nine had discontinued driving because of lack of funds (four patients), or after an illness (three patients) or accident (two patients). Eleven were unfit to drive because of visual defects (three patients), epilepsy (three patients), or severe cognitive or perceptual defects (three patients). Lack of confidence, particularly in wheelchair users, was a potent reason for giving up driving or not attempting to drive in the first place.

Employment

Eleven men and four women were in open employment. All of them had an IQ higher than 80. Five did clerical work, two were teachers, two were engineers, two were unskilled manual workers, and the others were a business executive, accountant, van driver, and builder. Three were studying in addition to working full time. Four men and six women were in sheltered employment.

Attainments

Twenty-eight survivors (49%) had one or more attainments in terms of living completely independently in the community (21 patients), driving a car (22 patients), or working in open employment (15 patients). Attainments were related to sensory level in infancy and to shunt history (Tables 1 and 3). Patients without a shunt or in whom the shunt was never revised were more likely to live independently, drive or work than those who needed revision, particularly after the age of two years when the cranial sutures had fused, rendering the intracranial contents more susceptible to pressure. Table 4 shows that late revisions of shunt (after the age of two years) were also associated with a birth head circumference greater than the 90th centile, a history of symptoms of raised intracranial pressure, visual defects, and need for daily care. Of the 11 survivors born with a birth head circumference greater than the 90th centile, nine needed revisions after the age of two years, ten had visual defects (of whom two were blind), seven had epilepsy, and five had an IQ lower than 80.

Discussion

Principal findings

By the mean age of 30 years, half the cohort had died and these were mainly the most disabled. One-third of the survivors lived independently, one-third needed some support and one-third needed daily care. Sensory level recorded in infancy predicted mortality, disability, and lifestyle in adulthood. Only about 10% of those who needed shunt revisions after the age of two years lived independently, drove a car, or worked in open employment.

Strengths and weaknesses of the study

The community basis of this study provides social as well as clinical data, enabling the realities of adulthood to be seen against the optimistic forecasts of the early years. Only 23 (40%) of the survivors were still attending hospital — mainly for urological care, pressure sores, or single items, such as surgical boots. Thus a hospital-based study would have given an incomplete perspective. As the patients grew older, the reduction in support, rehabilitation and encouragement from dedicated physiotherapists, parents and other carers revealed an outcome that was related to the patient's own motivation in addition to the basic neurological deficit.[8] A further strength of the study is that subjects were recruited consecutively without selection, and there was meticulous documentation of the clinical signs at birth.[4] Patients who never require a CSF shunt, or in whom the open lesion proves at operation to be a simple meningocele, have a better outlook and their proportion in any cohort is crucial to data on outcome. In the present series, there were 11 such patients, of

Table 2. Influence of sensory level and age on walking in 57 survivors with spina bifida.

Sensory level in infancy	All survivors (*n* = 57)	Walkers[a] at nine years (*n* = 32; 56%)	Walkers at 30 years (*n* = 17; 30%)
Above T11	13	0	0
T11–L3	16	5	0
L4	8	8	1
L5–S2	6	6	5
No sensory loss	11	11	10
Asymmetrical loss	3	2	1

[a]'Walkers' defined as able to walk more than 50 metres using aids if required. Survivors with lower sensory levels are more likely to be walkers — χ^2 for trend $P<0.0001$ for both age nine years and 30 years. Asymmetrical sensory loss excluded from the analysis.

Table 3. Lifestyle related to CSF shunt in 57 survivors at the mean age of 30 years.

	No shunt (*n* = 9)	Shunt,[a] no revisions (*n* = 16)	Shunt, revised at age <2 years (*n* = 10)	Shunt, revised at 2–30 years (*n* = 22)
Living independently, *n* = 21 (37%)	8	8	4	1[b]
Sheltered or in care	1	8	6	21
Driving a car, *n* = 22 (39%)	5	9	5	3[c]
Not driving	4	7	5	19
In open employment, *n* = 15 (26%)	3	6	4	2
Sheltered employment or none	6	10	6	20

[a]Shunts (*n* = 48) were only inserted or revised in response to definite clinical need, such as symptoms or signs of raised intracranial pressure. χ^2 for trend: [b]$P<0.0001$; [c]$P<0.05$.

Table 4. Features related to CSF shunt history in 57 patients with open spina bifida at the mean age of 30 years.

	No shunt n = 9	Shunt not revised n = 16	Shunt revised age <2 yrs n = 10	Shunt revised age 2 yrs n = 22	χ^2 for trend
Birth head circumference 90th centile, *n* = 11	0	1	1	9	$P = 0.01$
History of symptoms of raised intracranial pressure, *n* = 25	0	1	5	19	$P<0.0001$
Visual defects (mainly squint), *n* = 36	4	7	6	19	$P<0.05$
Daily care needed, *n* = 19	1	3	1	14	$P<0.01$

whom ten survived to age 30 years. In contrast, those born with a birth head circumference above the 90th centile had a much worse outcome, which may have been related to the effect of prenatal raised intracranial pressure or cortical maldevelopment.[9] The main limitation of the study is that the long follow-up implies treatments that have been superceded. Improvements in the diagnosis and management of neurological and renal problems have halved the mortality by the age of five years,[10,11] but have less influence on long-term disability.

Comparison with other studies

Outcome in childhood of early operated spina bifida has been widely reported,[1-5,11] but there are fewer studies of long-term outcome. A recent survey of a cohort of 118 adults aged 20 to 25 years with 24% mortality and 16% loss to follow-up, found continuing deterioration and a formidable number of neurosurgical and spinal operations.[12] This is the only 30-year prospective study of open spina bifida with 100% ascertainment by the same independent observer.

Implications

These data may help health professionals who counsel parents of children with spina bifida. They show a range of possible outcomes in adulthood when parents may no longer be able or willing to look after their child. The profound effects on the family,[13] and the implications of incontinence in the young adult must never be underestimated. Since, in this cohort, the most severely disabled died, modern treatment that reduces mortality may favour the survival of the more severely disabled.[6] Those caring for patients with spina bifida need to know both their long-term potential and the limited benefits of treatment to focus on realistic goals.[7]

The results have important practical implications for predicting long-term outcome in newborns. They suggest that babies with sensation below the knee (L3) are unlikely to be seriously disabled and could be achievers in adulthood. Babies who cry during a routine heel prick for the Guthrie test have a sensory level of S1 or below and are likely to remain community walkers at age 30. A similar response to pricking the saddle area (S2–4) forecasts the likelihood of bladder and bowel control.[14]

Such simple tests should be routine, and may be particularly useful for management decisions in countries with less access to specialist investigation and treatment.

In the UK, the care of children with spina bifida depends mainly on paediatricians. Once they are adults, patients lose that comprehensive care.[15] If they leave home they become separated from the support and care of parents whose experience of their condition is unique. The GP becomes the person most likely to be involved but may have limited experience of the particular problems presented by these patients. Chronic or intermittent shunt insufficiency has been misdiagnosed as sinusitis, dysphagia or painful neck, and referred to otolaryngologist, gastroenterologis or rheumatologist. Failure to recognise and treat shunt insufficiency promptly may be fatal, or may result in blindness or long-term dependency.[8,16,17] Although the incidence of spina bifida is falling, survivors from the 1960s and 1970s will continue to require long-term support.[15]

Conclusion

Survival in open spina bifida depends mainly on treatment. Disability and attainment depend on the severity of the original neurological deficit and on neurosurgical complications. A third of survivors to the age of 30 continue to need daily care.

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